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Enzyme therapy for pompe disease with recombinant human alpha-glucosidase from rabbit milk.

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Pompe disease is a metabolic myopathy caused by deficiency of lysosomal acid alpha-glucosidase. In this report we review the first 36 weeks of a clinical study on the safety and efficacy of enzyme therapy aimed at correcting the deficiency. Four patients with infantile Pompe disease were enrolled. They received recombinant human alpha-glucosidase from transgenic rabbit milk. The product is generally well tolerated and reaches the primary target tissues. Normalization of alpha-glucosidase activity in skeletal muscle was obtained and degradation of PAS-positive material was seen in tissue sections. The clinical condition of all patients improved. The effect on heart was most significant, with an impressive reduction of the left ventricular mass index (LVMI). Motor function improved. The positive preliminary results stimulate continuation and extension of efforts towards the realization of enzyme therapy for Pompe disease.

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- Clinical Trial

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1: Ann Neurol. 2004 Apr;55(4):495-502. Related Articles, Links

Enzyme replacement therapy in late-onset Pompe's disease: a three-year follow-up.

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Pompe's disease is an autosomal recessive myopathy. The characteristic lysosomal storage of glycogen is caused by acid alpha-glucosidase deficiency. Patients with late-onset Pompe's disease present with progressive muscle weakness also affecting pulmonary function. In search of a treatment, we investigated the feasibility of enzyme replacement therapy with recombinant human alpha-glucosidase from rabbit milk. Three patients (aged 11, 16, and 32 years) were enrolled in the study. They were all wheelchair-bound and two of them were ventilator dependent with a history of deteriorating pulmonary function. After 3 years of treatment with weekly infusions of alpha-glucosidase, the patients had stabilized pulmonary function and reported less fatigue. The youngest and least affected patient showed an impressive improvement of skeletal muscle strength and function. After 72 weeks of treatment, he could walk without support and finally abandoned his wheelchair. Our findings demonstrate that recombinant human alpha-glucosidase from rabbit milk has a therapeutic effect in late-onset Pompe's disease. There is good reason to continue the development of enzyme replacement therapy for Pompe's disease and to explore further the production of human therapeutic proteins in the milk of mammals.

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